OCA2 gene

OCA2 melanosomal transmembrane protein

Normal Function

The *OCA2* gene (formerly called the *P* gene) provides instructions for making a protein called the P protein. This protein is located in melanocytes, which are specialized cells that produce a pigment called melanin. Melanin is the substance that gives skin, hair, and eyes their color. Melanin is also found in the light-sensitive tissue at the back of the eye (the retina), where it plays a role in normal vision.

Although the exact function of the P protein is unknown, it is essential for normal pigmentation and is likely involved in the production of melanin. Within melanocytes, the P protein may transport molecules into and out of structures called melanosomes (where melanin is produced). Researchers believe that this protein may also help regulate the relative acidity (pH) of melanosomes. Tight control of pH is necessary for most biological processes.

Health Conditions Related to Genetic Changes

Angelman syndrome

The *OCA2* gene is located in a region of chromosome 15 that is often deleted in individuals with Angelman syndrome. A loss of this gene does not cause the characteristic neurologic features of Angelman syndrome; however, people with this condition who are missing one copy of the *OCA2* gene tend to have unusually light-colored hair and fair skin. Cells with only one copy of the *OCA2* gene make a reduced amount of P protein compared with cells with two functional copies of this gene, which affects the coloring of the hair and skin.

A small percentage of people with Angelman syndrome also have oculocutaneous albinism type 2. This condition occurs when people have two nonfunctional copies of the *OCA2* gene in each cell. In addition to a deletion in chromosome 15 that removes one copy of the *OCA2* gene, these individuals have a mutation in the *OCA2* gene on the other copy of chromosome 15. As a result, cells make little or no functional P protein. A lack of P protein disrupts the production of melanin, leading to the characteristic features of albinism.

oculocutaneous albinism

More than 80 mutations in the *OCA2* gene have been identified in people with oculocutaneous albinism type 2. People with this form of albinism often have light yellow, blond, or light brown hair; creamy white skin; light-colored eyes; and problems

with vision. The most common *OCA2* mutation is a large deletion in the gene, which is found in many affected individuals of sub-Saharan African heritage. Other *OCA2* gene mutations, including changes in single DNA building blocks (base pairs) and small deletions, are more common in other populations. Mutations in the *OCA2* gene disrupt the normal production of melanin, which reduces coloring of the hair, skin, and eyes and affects vision.

Prader-Willi syndrome

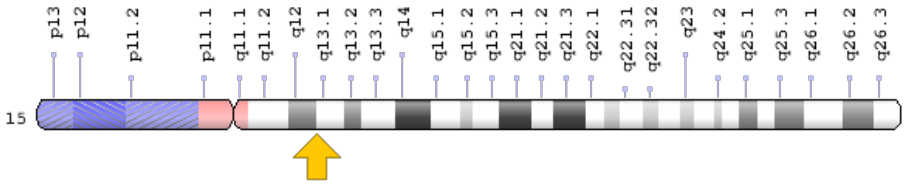
The region of chromosome 15 containing the *OCA2* gene is often deleted in individuals with Prader-Willi syndrome. A loss of this gene does not cause intellectual disability and the other characteristic features of Prader-Willi syndrome; however, people with this condition who are missing one copy of the *OCA2* gene tend to have unusually light-colored hair and fair skin. Cells missing a copy of the *OCA2* gene make less P protein than cells with two functional copies of the gene, which affects the coloring of the hair and skin.

Oculocutaneous albinism type 2 also occurs in a small number of people with Prader-Willi syndrome. This condition occurs when people have two nonfunctional copies of the *OCA2* gene in each cell. In addition to a deletion in chromosome 15 that removes one copy of the *OCA2* gene, these individuals have a mutation in the *OCA2* gene on the other copy of chromosome 15. As a result, cells make little or no functional P protein. A lack of P protein disrupts the production of melanin, leading to the characteristic features of albinism.

Chromosomal Location

Cytogenetic Location: 15q12-q13.1, which is the long (q) arm of chromosome 15 between positions 12 and 13.1

Molecular Location: base pairs 27,719,008 to 28,099,342 on chromosome 15 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- BOCA
- Melanocyte-specific transporter protein
- oculocutaneous albinism II
- oculocutaneous albinism II (pink-eye dilution homolog, mouse)
- P gene
- P_HUMAN
- PED
- Pink-eyed dilution protein homolog

Additional Information & Resources

Educational Resources

- Protein Spotlight, Swiss Institute of Bioinformatics
  http://web.expasy.org/spotlight/back_issues/054/

GeneReviews

- Angelman Syndrome
  https://www.ncbi.nlm.nih.gov/books/NBK1144
- Oculocutaneous Albinism Type 2
  https://www.ncbi.nlm.nih.gov/books/NBK1232
- Prader-Willi Syndrome
  https://www.ncbi.nlm.nih.gov/books/NBK1330

Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28OCA2%5BTIAB%5D%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1800+days%22%5Bdp%5D

OMIM

- OCA2 GENE
  http://omim.org/entry/611409

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
  http://atlasgeneticsoncology.org/Genes/OCA2ID45789ch15q12.html
- ClinVar
  https://www.ncbi.nlm.nih.gov/clinvar?term=OCA2%5Bgene%5D
- HGNC Gene Symbol Report
  http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=8101
- International Albinism Center, University of Minnesota: Albinism Database
  http://www.ifpcs.org/albinism/oca2mut.html
- NCBI Gene
  https://www.ncbi.nlm.nih.gov/gene/4948
- UniProt
  http://www.uniprot.org/uniprot/Q04671

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